PHKG2 gene

phosphorylase kinase catalytic subunit gamma 2

Normal Function

The *PHKG2* gene provides instructions for making one piece, the gamma subunit, of the phosphorylase b kinase enzyme. This enzyme is made up of 16 subunits, four each of the alpha, beta, gamma, and delta subunits. (Each subunit is produced from a different gene.) The gamma subunit performs the function of phosphorylase b kinase enzyme, and the other subunits help regulate its activity. This enzyme is found in various tissues, although it is most abundant in the liver and muscles. One version of the enzyme is found in liver cells and another in muscle cells. The gamma-2 subunit produced from the *PHKG2* gene is part of the enzyme found in the liver.

Phosphorylase b kinase plays an important role in providing energy for cells. The main source of cellular energy is a simple sugar called glucose. Glucose is stored in muscle and liver cells in a form called glycogen. Glycogen can be broken down rapidly when glucose is needed, for instance to maintain normal levels of glucose in the blood between meals. Phosphorylase b kinase turns on (activates) another enzyme called glycogen phosphorylase b by converting it to the more active form, glycogen phosphorylase a. When active, this enzyme breaks down glycogen.

Health Conditions Related to Genetic Changes

glycogen storage disease type IX

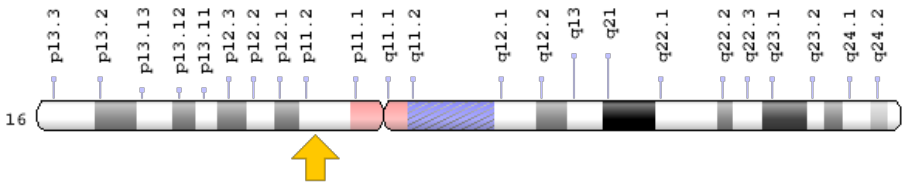
At least 30 mutations in the *PHKG2* gene are known to cause a form of glycogen storage disease type IX (GSD IX) called GSD IXc. This form of the disorder affects liver function and causes an enlarged liver (hepatomegaly), slow growth, and periods of low blood sugar (hypoglycemia) in affected individuals. These features usually improve over time. However, *PHKG2* gene mutations have been associated with more severe signs and symptoms such as irreversible liver disease (cirrhosis).

Mutations in the *PHKG2* gene reduce the activity of phosphorylase b kinase in liver cells, although the mechanism is unknown. Reduction of this enzyme's function impairs glycogen breakdown. As a result, glycogen builds up in cells, and glucose is not available for energy. The inability to break down glycogen in the liver causes the features of GSD IXc.

Chromosomal Location

Cytogenetic Location: 16p11.2, which is the short (p) arm of chromosome 16 at position 11.2

Molecular Location: base pairs 30,748,299 to 30,761,176 on chromosome 16 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- GSD9C
- PHK-gamma-T
- PHKG2_HUMAN
- phosphorylase b kinase gamma catalytic chain, testis/liver isoform
- phosphorylase kinase gamma subunit 2
- phosphorylase kinase subunit gamma-2
- phosphorylase kinase, gamma 2 (testis)
- phosphorylase kinase, gamma 2 (testis/liver)
- PSK-C3
- serine/threonine-protein kinase PHKG2

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): Glycogen Metabolism
  https://www.ncbi.nlm.nih.gov/books/NBK21190/

GeneReviews

- Phosphorylase Kinase Deficiency
  https://www.ncbi.nlm.nih.gov/books/NBK55061

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28PHKG2%5BTIAB%5D%29+OR+%28%28PHK-gamma-T%5BTIAB%5D%29+OR+%28PSK-C3%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- PHOSPHORYLASE KINASE, TESTIS/LIVER, GAMMA-2
  http://omim.org/entry/172471

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_PHKG2.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=PHKG2%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=8931
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/5261
- UniProt
  http://www.uniprot.org/uniprot/P15735

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